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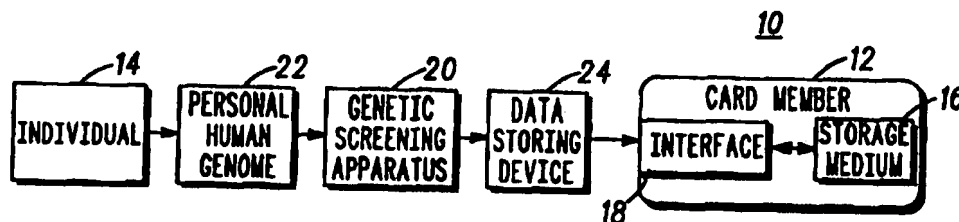
(57) Abstract

A personal genome card (10) includes a card member (12) and a machine-readable storage medium (16) integrated in the card member. Machine-readable data including a representation of a sequence of nucleotide bases for at least a portion of a genome of an individual (22) is stored on the machine-readable storage medium (16). The machine-readable data can further include phenotype information for the individual, a personal medical history of the individual, and genetic pedigree information. Optionally, a processor is integrated in the card member. The processor is operative to limit access to at least a portion of the information stored in the personal genome card (10).

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PERSONAL HUMAN GENOME CARD AND METHODS
AND SYSTEMS FOR PRODUCING SAME

15   Field of the Invention

The present invention relates to personal medical
information cards.

20   Background of the Invention

The set of instructions for making an organism is
called its genome. The genome for a human consists of
coiled threads of deoxyribonucleic acid (DNA) and
25   associated protein molecules found in every cell nucleus
of the human. The components of these threads of DNA
encode all of the information needed for building and
maintaining life in the human.
        Each DNA molecule contains a number of genes, which
30   are the basic physical and functional units of heredity.
A gene is a specific sequence of nucleotide bases within
the human genome. The human genome is estimated to
comprise approximately 50,000 to 100,000 genes. A
number of these genes, estimated at 4000, are involved
35   in an individual's predisposition to disease. The Human

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Genome Project was initiated in 1990 with a goal of
characterizing the entire human genome in order to
devise improved strategies for disease identification,
treatment, and prevention.

5   The prospect of determining an individual's
predisposition to disease based on his/her genome has
led some to warn of the potential for genetic-based
discrimination in health insurance coverage. Here,
genetic information could be used to exclude high-risk
10   individuals from health care by either denying coverage
or by charging prohibitive rates. The fear of
discrimination may act to inhibit an individual from
divulging his/her genetic status, even in light of the
benefits of preventative measures and treatments which
15   can be prescribed based on an early diagnosis of genetic
predisposition to disease.

                    Brief Description of the Drawings

20   The invention is pointed out with particularity in
the appended claims. However, other features of the
invention will become more apparent and the invention
will be best understood by referring to the following
detailed description in conjunction with the
25   accompanying drawings in which:
        FIG. 1 is a block diagram of a system for producing
a personal genome card in accordance with the present
invention;
        FIG. 2 is a flow chart of an embodiment of a method
30   of producing a personal genome card for an individual;
        FIG. 3 is a block diagram illustrating a system for
storing information in a personal genome card;
        FIG. 4 is an illustration of a PCMCIA embodiment of
a personal genome card;

FIG. 5 is a block diagram of a smart card embodiment of a personal genome card; and

FIG. 6 is a block diagram illustrating a system for accessing a personal genome card.

Detailed Description of a Preferred Embodiment

Embodiments of the present invention advantageously provide a personal genome card containing a stored representation of an individual's genome. The personal genome card is sized to be carried by the individual for use in both medical and personal identification applications. Embodiments of the personal genome card limit external access to at least a portion of the representation of the individual's genome in order to avoid instances of genetic-based discrimination.

FIG. 1 is a block diagram of a system for producing a personal genome card 10 in accordance with the present invention. The personal genome card 10 includes a card member 12 which, preferably, is sized for carrying on an individual 14. For example, the card member 12 can be the size of a credit card for carrying within a wallet, a purse, or a pocket of the individual 14.

A machine-readable storage medium 16 is integrated in the card member 12. The machine-readable storage medium 16 is utilized to store personal genome information for the individual 14. The machine-readable storage medium 16 can be in the form of an electronic memory device, a magnetic memory device, or an optical memory device, for example. Preferably, the machine-readable storage medium 16 is non-volatile so that a powering signal need not be applied to maintain the personal genome information therein. Further, it is preferred that the machine-readable storage medium 16 be

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rewriteable so that the information contained therein
can be updated.

Also integrated within the card member 12 is an
interface 18. The interface 18 is utilized to
5   communicate the personal genome information between the
machine-readable storage medium 16 and an exterior
read/write device such as a computer.

A genetic screening apparatus 20 is utilized to
screen at least a portion of a personal human genome 22
10 of the individual 14. The genetic screening apparatus
20 forms the personal genome information to be stored in
the personal genome card 10. The personal genome
information is communicated to a data storing device 24
which performs a step of storing a representation
15 thereof in the machine-readable storage medium 16. The
data storing device 24 can be embodied a computer, for
example, which communicates the personal genome
information to the machine-readable storage medium 16
via the interface 18. In one embodiment, the personal
20 genome card 10 includes a PC card, such as a PCMCIA
card, having an internal memory for storing the personal
genome information. Here, the interface 18 includes a
PC card interface which is received by a mating PC card
interface in the computer.

25   The personal genome information produced by the
genetic screening apparatus 20 can be in any of a
variety of forms. In one form, the genetic screening
apparatus 20 determines a sequence of nucleotide bases
for at least a portion of the personal human genome 22
30 of the individual 14. The sequence of nucleotide bases
can be determined from either a DNA sample or an RNA
sample of the individual 14. The DNA or RNA sample can
be sequenced to determine either a partial nucleotide

sequence or an entire nucleotide sequence of the
personal human genome 22 of the individual 14.

Further, a sequence of nucleotide bases can be
determined from a messenger RNA (mRNA) sample from the
5   individual 14, or equivalently from copy DNA (cDNA)
synthesized from the mRNA sample. The sequence of
nucleotide bases in mRNA or cDNA is indicative of a
protein-coding portion of the DNA, i.e. a gene of the
individual 14. The existence of mRNA provides evidence
10 of an active or expressed gene within the individual 14.

In these embodiments, the data storing device 24
stores a representation of the sequence of nucleotide
bases, determined from any of the above-described
approaches, in the machine-readable storage medium 16.
15 For a DNA sample, the nucleotide bases within the
sequence are selected from adenine (A), cytosine (C),
guanine (G), and thymine (T). For an RNA sample, the
nucleotide bases within the sequence are selected from
adenine (A), cytosine (C), guanine (G), and uracil (U).
20 Regardless of whether a DNA or an RNA sample is
sequenced, the representation can include an
uncompressed sequence of two-bit codes, wherein each
two-bit code indicates one of four different nucleotide
bases which comprise the sequence. Alternatively, the
25 representation can include a lossless, compressed
representation of the sequence. Various lossless data
compression techniques can be utilized for this purpose.

Another form of personal genome information which
can be determined by the genetic screening apparatus 20
30 is a genetic map for the individual 14. The genetic map
is representative of positions of predetermined
molecular markers within the DNA or RNA of the
individual 14. The molecular markers can be determined
using a restriction enzyme which cuts the DNA or the RNA

into fragments about a predetermined nucleotide subsequence. For example, a restriction enzyme can be utilized to determine the position of a molecular marker in the form of a G-A-T-C subsequence.

5   Using this form of personal genome information, the data storing device 24 stores a representation of the genetic map in the machine-readable storage medium 16. The representation can include a sequence of codes representative of distances between adjacent molecular

10   markers. Alternatively, the representation can include a sequence of codes representative of absolute positions of the molecular markers. A lossless data compression technique can be utilized to produce a lossless, compressed representation based on either approach for

15   storing the genetic map.

Regardless of its form, the personal genome information stored in the machine-readable storage medium 16 either directly or indirectly indicates if the individual 14 has a genetically-inherited disease and/or

20   a genetic predisposition to a disease. If so, an appropriate treatment strategy can be formulated by medical personnel.

The foregoing description of preferred embodiments of the present invention is based on a nucleotide

25   sequence form for the personal genome information. It is noted, however, that alternative embodiments may utilize other forms for the personal genome information, such as a genetic map form.

FIG. 2 is a flow chart of an embodiment of a method

30   of producing a personal genome card for an individual. The method includes a step of providing the card member 12 having the machine-readable storage medium 16 integrated therein, as indicated by block 40. A smart

card or a PC card, such as a PCMCIA card, can be
utilized to provide the card member 12.

As indicated by block 42, a step of determining a
first sequence of nucleotide bases for at least a

5   portion of a genome of the individual 14 is performed.
The first sequence of nucleotide bases can be determined
by a genetic sequencing apparatus in the genetic
screening apparatus 20. If desired, the sequence of
nucleotide bases can be determined for an entire exon,

10 for an entire gene, for an entire chromosome, or for the
entire genome of the individual 14.

A step of storing a representation of the first
sequence in the machine-readable storage medium 16 is
performed, as indicated by block 44. The step of

15 storing the representation of the first sequence can
include compressing the first sequence to form lossless,
compressed data, and storing the lossless, compressed
data in the machine-readable storage medium. If the
first sequence includes an exon or a gene having a

20 predetermined sequence, the lossless, compressed data
can include a code which identifies the predetermined
sequence for the exon or the gene. For example, if the
exon or the gene has a sequence observed in other
individuals, a single code can be utilized to compress

25 the representation of the sequence.

Alternatively, if the first sequence includes an
exon or a gene which is either a corrupted or a mutated
version of a predetermined sequence, the lossless,
compressed data can include a first code identifying the

30 predetermined sequence and a second code indicating the
corruption or the mutation. The corruption or the
mutation can be present at the birth of the individual
14 or can be incident-based, i.e. an aberration from the
individual's genes at birth.

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As indicated by block 46, the method can include a
step of storing, in the machine-readable storage medium
16, phenotype information for the individual 14.
Preferably, the phenotype information is associated with
5   genotype information encoded by the first sequence. The
method can further include a step of storing, in the
machine-readable storage medium 16, a personal medical
history for the individual 14, as indicated by block 48.
      Optionally, the method can include steps of storing
10   genome information and medical information for one or
more family members of the individual 14. In
particular, a step of determining a second sequence of
nucleotide bases for at least a portion of a genome of a
family member of the individual 14 is performed, as
15   indicated by block 50. A step of storing a
representation of the second sequence of nucleotide
bases in the machine-readable storage medium 16 is
performed, as indicated by block 52. As indicated by
block 54, a step of storing, in the machine-readable
20   storage medium 16, phenotype information for the family
member is performed. Further, a step of storing a
personal medical history for the family member can be
performed as indicated by block 56.
      The steps indicated by blocks 50, 52, 54, and 56
25   can be repeated for a plurality of family members of the
individual 14. Preferably, the plurality of family
members include at least one ancestor of the individual
14, such as a mother and/or a father of the individual
14. By including genor   nformation and medical history
30   for at least one ances1   of the individual 14, the
personal genome card 1(ntains genetic pedigree
information for the ind. idual 14. The genetic pedigree
information is useful for predicting diseases whose
onset are probabilistic for individuals having a

predetermined disease gene. For example, if a disease
gene is identified in the genome of the individual 14,
the genetic pedigree information can be referred to
determine if ancestors of the individual 14 having the
5   same disease gene expressed a corresponding disease.
FIG. 3 is a block diagram illustrating a system for
storing information in a personal genome card 60. A
computer 62 or like programmable apparatus receives or
determines a first sequence of nucleotide bases 64 for
10   the individual 14, phenotype information 66 for the
individual 14, a personal medical history 68 for the
individual 14, a second sequence of nucleotide bases 70
for at least one family member of the individual 14,
phenotype information 72 for the family member, a
15   personal medical history 74 for the family member, and
identification information 76 (such as name, social
security number, or insurance number) for the individual
14. The computer 62 stores this information in a
storage medium 78 interfaced to the computer 62 via an
20   interface 80. The system described is amenable to a
PCMCIA embodiment of the personal genome card 60,
wherein the interface 80 is a PCMCIA interface.
FIG. 4 is an illustration of a PCMCIA embodiment of
a personal genome card 90. The personal genome card 90
25   includes a card member 92 having a PCMCIA interface 94.
A storage medium (not illustrated) such as a memory is
integrated within the card member 92. Here, the
personal genome card 90 can be received in a PCMCIA port
on the computer 62 for storing personal genome
30   information, personal medical history, and pedigree
information in the memory.
FIG. 5 is a block diagram of a smart card
embodiment of a personal genome card 100. Similar to
the embodiment of FIG. 1, the personal genome card 100

includes a machine-readable storage medium 102 and an
interface 104 integrated within a card member 106.

In addition, a processor 108 is integrated in the
card member 106. The processor 108 is operative to
5   limit external access to predetermined information
stored in the machine-readable storage medium 102. The
access is allowed or denied based upon whether a
predetermined access code is provided to the processor
108. The predetermined access code can be a secret code
10   known only by the individual 14. The processor 108
receives the predetermined access code via the interface
104 or via a user interface 109 incorporated in the card
member 106. Having the user interface 109 (such as a
series of buttons) incorporated in the card member 106
15   allows the individual 14 to enter the predetermined
access code without having to divulge the predetermined
access code to others.

Preferably, the processor 108 does not limit
external access to personal medical information which
20   would be vital in an emergency medical situation where
communication with the individual 14 is either impaired
or impossible. However, the processor 108 does limit
external access to at least a portion of the personal
genome information. This access is limited to avoid a
25   possibility of discrimination in obtaining health
insurance or discrimination in hiring based on having a
genetic predisposition to a disease.

The processor 108 can also be operative to
decompress the lossless, compressed data stored on the
30   machine-readable storage medium 102. As a result,
decompressed data is communicated via the interface 104.

To produce the smart card embodiment of the
personal genome card, a step of integrating the
processor 108 in the card member 106 is performed in

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addition to the steps illustrated in FIG. 2. This step
can include storing the predetermined access code and
access software either in the processor 108 or the
machine-readable storage medium 102.

5   FIG. 6 is a block diagram illustrating a system for
accessing a personal genome card 110. The personal
genome card 110 communicates with an on-line system 112.
The on-line system 112 accesses the information stored
in a machine-readable storage medium 111 for analysis
10   and/or display. The on-line system 112 can be located
remotely from the personal genome card 110, as
illustrated in FIG. 6, or can be co-located with the
personal genome card 110.

        The personal genome card 110 is received by an
15   interface 114. The interface 114 mates with an
interface 116 within the personal genome card 110. For
remote access, a modem 118 is coupled to the interface
114 to communicate data with a remote modem 120. The
modem 118 can communicate with the remote modem 120
20   using either a wireline link or a wireless link. The
remote modem 120 communicates the data to a computer
system having a computer 122, a user interface 124 (e.g.
a keyboard and/or a mouse), and a display device 126
(e.g. a computer monitor). If the personal genome card
25   110 is co-located with the on-line system 112, the modem
118 and the remote modem 120 are not required.

        The on-line system 112 can be utilized by medical
personnel to diagnose the individual's genetic
condition. The medical personnel can navigate the
30   information stored the personal genome card 110 by
entering navigation instructions into the user interface
124. The navigation instructions are communicated to
the personal genome card 110 by the remote modem 120,
the modem 118, and the interface 114. In response

thereto, the personal genome card 110 communicates
either compressed data or uncompressed data to the
computer 122 by the interface 114, the modem 118, and
the remote modem 120. If compressed data is
5   transmitted, the computer 122 includes decompression
software to decompress the personal genome information.
As an alternative to remotely controlling which of the
personal genome information is transmitted, a
predetermined amount of the data can be transmitted to
10   the computer 122.

The computer 122 is utilized for analysis and/or
display of the personal genome information, the personal
medical history, and the pedigree information. Medical
personnel can use the analysis of the personal genome
15   information to determine if the individual 14 has a
genetically-inherited disease or a genetic
predisposition to a disease. Further, a doctor can
compare the personal genome information in the personal
genome card 110 to more-recently screened information to
20   determine if any mutations have occurred since a
previous screening. The personal medical history can be
utilized to determine if the individual 14 has
previously exhibited any symptoms of the disease. The
pedigree information can be utilized to gauge the
25   likelihood of an onset of the disease based on the
genome and medical history of the individual's
ancestors. The personal genome information is also
useful in prescribing drugs which will not adversely
affect the individual 14.

30   The on-line system 112 can also be utilized for
remote identification purposes. Here, the on-line
system 112 contains a database 130 of personal genome
information for a plurality of individuals, and compares
the personal genome information received from the

personal genome card 110 thereto. The on-line system
112 can also be utilized to store updates of an
individual's genome over his/her lifetime.

   Thus, there has been described herein a concept, as
5 well as several embodiments including preferred
embodiments of a personal human genome card and a method
and system of producing the same.

   Because the various embodiments of the present
invention store personal genome information in a card
10 which can be carried by an individual, they provide a
significant improvement in that the card can be used by
medical personnel to determine if the individual has a
genetic predisposition to disease. As more disease
genes are identified in scientific research, the
15 personal genome information in the personal genome card
can be reviewed as a preventative diagnostic procedure.
As a result of the preventative diagnostic, medical
personnel may prescribe preventative measures for the
individual to reduce a likelihood of acquiring a
20 disease.

   Additionally, the various embodiments of the
present invention as herein-described limit external
access to at least a portion of the personal genome
information in order to avoid genetic-based
25 discrimination.

   It will be apparent to those skilled in the art
that the disclosed invention may be modified in numerous
ways and may assume many embodiments other than the
preferred form specifically set out and described above.
30 Accordingly, it is intended by the appended claims
to cover all modifications of the invention which fall
within the true spirit and scope of the invention.

   What is claimed is:

Claims

1.   A method of producing a personal genome card
for an individual, the method comprising the steps of:
5   providing a card member having a machine-readable
storage medium integrated therein, the card member sized
for carrying on the individual;
         determining a first sequence of nucleotide bases
for at least a portion of a genome of the individual;
10   and
         storing a representation of the first sequence in
the machine-readable storage medium.

2.   The method of claim 1 wherein the step of
15   storing the representation of the first sequence
includes:
         compressing the first sequence to form lossless,
compressed data; and
         storing the lossless, compressed data in the
20   machine-readable storage medium.

3.   The method of claim 1 further comprising the
step of storing phenotype information for the individual
on the machine-readable storage medium.
25

4.   The method of claim 1 further comprising the
steps of:
         determining a second sequence of nucleotide bases
for at least a portion of a genome of an ancestor of the
30   individual; and
         storing a representation of the second sequence in
the machine-readable storage medium.

5.   A personal genome card for an individual, the personal genome card comprising:

a card member sized for carrying on the individual;

a machine-readable storage medium integrated in the

5   card member; and

machine-readable data stored on the machine-readable storage medium, the machine-readable data including a representation of a first sequence of nucleotide bases for at least a portion of a genome of

10   the individual.

6.   The personal genome card of claim 5 wherein the first sequence includes an entire nucleotide sequence of the genome of the individual.

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7.   The personal genome card of claim 5 wherein the representation includes lossless, compressed data for the first sequence.

20   8.   The personal genome card of claim 7 wherein the first sequence includes a gene having a predetermined sequence, and wherein the lossless, compressed data includes a code which identifies the predetermined sequence for the gene.

25

9.   The personal genome card of claim 5 wherein the machine-readable data further includes phenotype information for the individual.

30   10.   The personal genome card of claim 5 wherein the machine-readable data further includes a representation of a second sequence of nucleotide bases for at least a portion of a genome of an ancestor of the individual.
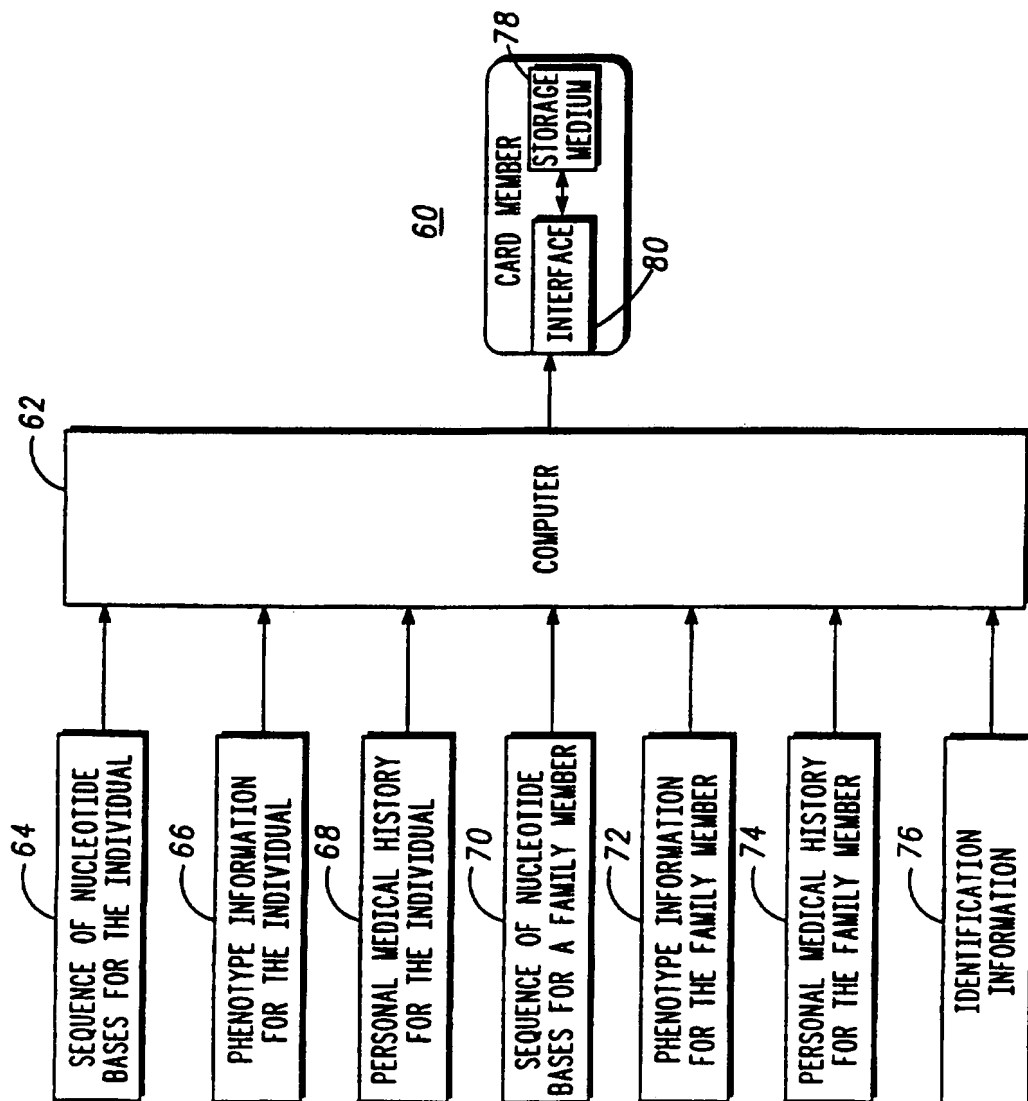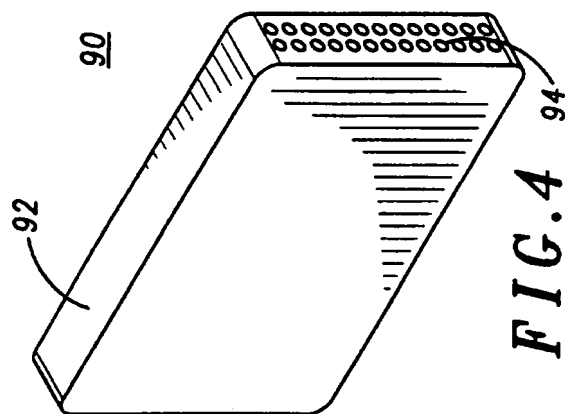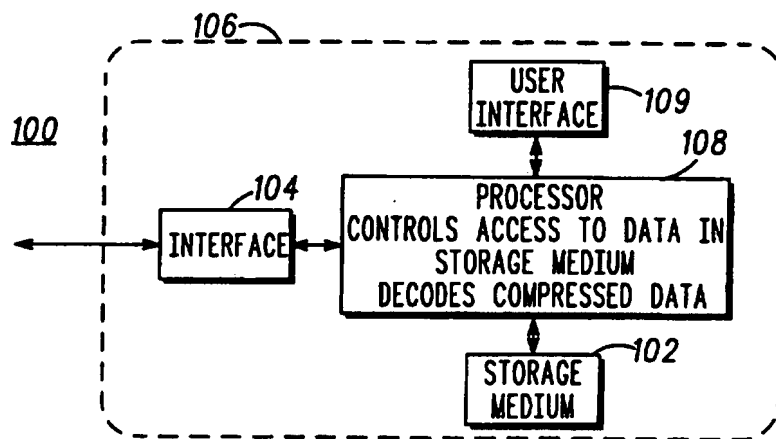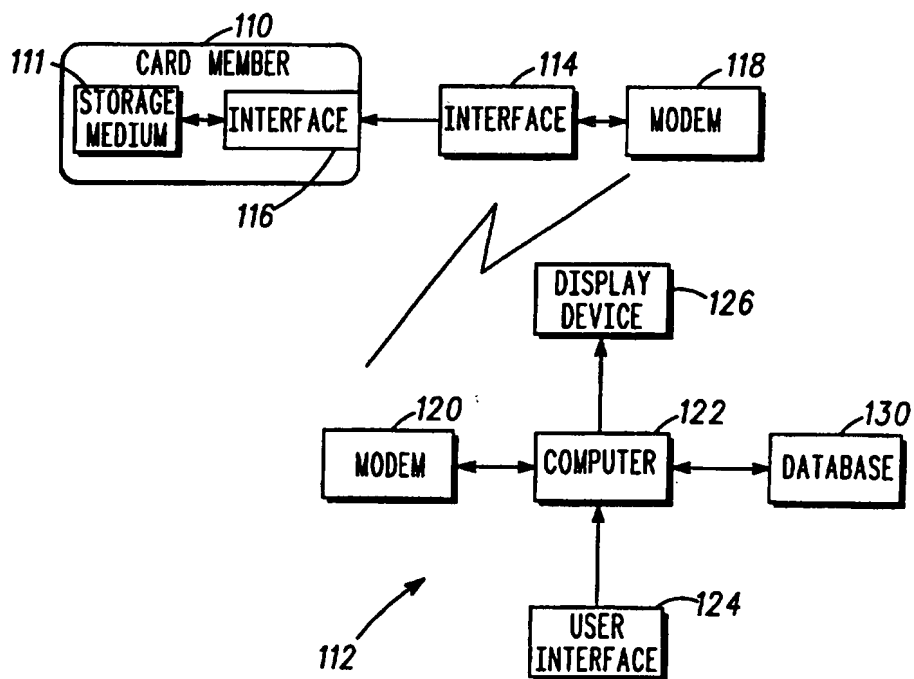
FIG.4

FIG.3

FIG.5

FIG.6

INTERNATIONAL SEARCH REPORT

A. CLASSIFICATION OF SUBJECT MATTER

IPC(6) :G06K 7/08

US CL : 364/496; 235/492;

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

U.S. : 364/413.01, 413.02, 496-498; 235/492

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practicable, search terms used)

APS, DIALOG

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
Y	US 5,473,690 (GRIMONPREZ et al) 05 December 1995, col. 1, lines 7-23.	1-10.
Y	US 5,196,682 (ENGLEHARDT) 23 March 1993, col. 8, lines 33-43.	1-10.
Y	US 4,891,506 (YOSHIMATSU) 02 January 1990, col. 3, line 62- col. 4, line 14.	1-10.
Y	US 5,371,671 (ANDERSEN et al) 06 December 1994, col. 2, lines 2-15, 36-42.	1-10.
Y	US 4,982,326 (KANEKO) 01 January 1991, col. 13, lines 50-56.	1-10.
Y	US 4,802,101 (HARA) 31 January 1989, col. 8, lines 8-26.	1-10.

[X] Further documents are listed in the continuation of Box C.   [ ] See patent family annex.

	Special categories of cited documents:		
"A"	document defining the general state of the art which is not considered to be part of particular relevance	"T"	later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention
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"P"	document published prior to the international filing date but later than the priority date claimed	"&"	document member of the same patent family

Date of the actual completion of the international search

20 MARCH 1997

Date of mailing of the international search report

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C (Continuation). DOCUMENTS CONSIDERED TO BE RELEVANT		
Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	US 5,291,399 (CHACO) 01 March 1994.	1-10.
A	US 4,852,570 (LEVINE) 01 August 1989.	1-10.